American Speech-Language-Hearing Association

LET's TALK

...FOR PEOPLE WITH SPECIAL COMMUNICATION NEEDS

Genetic Hearing Loss and Genetic Counseling

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What is a "gene"?

Genes come in pairs, and we receive one gene of every pair from each of our parents. Genes are written in a code called DNA, which instructs the body to produce specific chemicals. When a gene does not work properly, it does not produce the chemicals correctly. This can lead to problems in the body's structure, function, and/or appearance.

How is hearing loss inherited?

Some genetic hearing losses only happen when the same affected gene is inherited from both the mother and the father. This is called "**recessive**." Other genetic hearing losses can happen when only one parent has the affected gene. This is called "**dominant**."

Other times, the parents both have normal genes, but an alteration or mutation happens in the sperm or egg cell that formed the baby. Once the alteration happens in one cell, it is duplicated in all of the cells in the body. Therefore, genetic hearing loss can happen even if there are no other family members who have hearing loss.

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There are other categories of genetic hearing loss, including "complex" causes. An example of a complex cause is when certain people have a gene that makes them more likely to develop a hearing loss after taking antibiotics.

Is hearing loss always seen by itself?

Sometimes a genetic hearing loss happens with other health problems, such as kidney problems or vision loss. When hearing loss occurs with other health problems, it is called "syndromic" hearing loss.

When there is a hearing loss but no other obvious health problems that can be grouped together in a syndrome, it is called "**nonsyndromic**" hearing loss. A common cause of nonsyndromic hearing loss is a change in a gene called GJB2. This disorder may also be referred to as Connexin26 or DFNB1.

Sometimes symptoms of other health problems may not show up until later in life. If this happens, the child may appear to be nonsyndromic at first, and then syndromic later in life.

Why would I want to know if my child's hearing loss is genetic?

If the hearing loss has a genetic cause, there are two important issues to consider. First, your other children (or future children) may be at greater risk to have a hearing loss as well. They should have a hearing test as soon as possible, and their hearing should be monitored. Second, there may be other health issues to address if the hearing loss is part of a syndrome.

How do I find out if my child's hearing loss has a genetic cause?

A genetic counselor can help you determine the cause of the hearing loss (if possible), educate you and your family about the condition, and refer you to other health care professionals if necessary.

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Where do I find a genetic counselor who specializes in hearing loss?

Your child's audiologist or physician can refer you to a genetic counselor, or you can find a genetic counselor at the National Society of Genetic Counselors Web site (**www.nsgc.org**).

You might see only a genetic counselor, or you might see a genetic counselor with a team of health professionals. A multidisciplinary team may include a genetic counselor, an ear-nose-and-throat physician (ENT), a geneticist, and an audiologist. Geneticists can be medical doctors (MDs) or research doctors (PhDs).

How do I prepare for a genetic evaluation?

The genetic counselor will construct a "pedigree," or family tree that contains information about which family members have hearing loss or other health issues of interest. As you speak with your family, try to find out as much as you can. Ask questions such as:

- "Did the hearing loss begin in childhood?"
- "What was the degree of the hearing loss?"
- "Did the hearing loss get worse over time?"

In learning more about your family history, your family members may find out that your child has a hearing loss with a possible genetic cause.

The genetic counselor also will obtain a "**case history**." The counselor will ask many questions. Some will be related to genetics and hearing loss, others will not. The genetic counselor will help you find out whether the hearing loss is genetic or due to another factor. Having as much of the following information as possible will assist the genetic counselor. In turn, the genetic counselor will be able to give your family more complete answers.

- Your child's prenatal, delivery, and newborn history, especially any complications, illnesses, or other factors you think might be important
- Developmental milestones
- Medical history
- School progress
- Hearing history: Has your child's hearing changed? When did you first suspect a hearing loss and why?

In learning more about your family history, your family members may find out that your child has a hearing loss with a possible genetic cause.

Make sure you have copies of results from tests such as the Newborn Hearing Screen, ENT examinations, hearing tests, or CT scans. You may need to ask your health care providers to send copies of the results to the genetic counselor.

It is very important that you have all of this information available for the genetic evaluation so that appropriate recommendations can be made, and to avoid delays in the referral process if other tests are needed. In some cases (e.g., foster care, adoption), this information may not be available. Just do the best that you can.

What should I expect from a genetic evaluation?

You may have one or two visits, depending upon whether further testing is recommended. In a team evaluation, the first visit usually involves a review of the case history (described above), a physical examination, a hearing test, a review of other test results, a discussion of impressions, and recommendations.

During the physical examination, the physician will look for subtle differences that are not noticeable to the average person. For instance, the number and color of birthmarks, the color of the eyes, the shape and position of the ears, tiny indentations or pieces of skin around the ear and neck, and the facial appearance will be noted.

Although your child may have had a hearing test already, the team (or your child's audiologist) may recommend further testing to obtain more complete information, and to determine whether the hearing loss is stable. In order to make a diagnosis, the team needs to know the following:

- The severity of the loss
- Whether the hearing loss gets worse over time (progressive)

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- Whether the hearing loss was present at birth (congenital) or developed later (acquired)
- Whether the hearing loss is in both ears (bilateral) or one ear (unilateral)
- Whether the loss is due to inner ear or nerve dysfunction (sensorineural), external or middle ear dysfunction (conductive), or a combination of problems (mixed)

Other tests that might be recommended (if not already done) include (a) a head CT scan to look for irregularities in the structures of the middle and inner ears; (b) a vision test because children with hearing loss may rely more on visual cues, and because certain syndromes involve vision loss; (c) balance, heart, or kidney tests if there is a reason to suspect a genetic disorder that involves these organs; and (d) other diagnostic blood tests to look for infections or other problems that might rule out a genetic cause. A blood test can also find certain types of gene abnormalities, such as GJB2. Depending on the laboratory that does the tests and how complicated the test is, it may take several months to obtain results.

Will insurance pay for all or part of the recommended tests?

Fees will be billed for the office visits and laboratory tests. Some insurance companies will not cover genetic testing, even though it is considered diagnostic. You should check with your insurance company before you begin the process to determine the level of coverage. The genetic counselor may be able to help if you experience difficulty.

How do I learn more?

When you are ready to learn more about genetic hearing loss and counseling, ask your audiologist for a list of additional references.

Information for CD jacket:

Special thanks to Angelique Boerst for helpful comments and suggestions on previous drafts of this patient information sheet. Ms. Boerst is an audiologist at the University of Michigan.

Additional resources:

www.handsandvoices.org

www.babyhearing.org

www.agbell.org

www.hereditaryhearingloss.org

For more information about hearing loss, hearing aids or referral to an ASHA-certified audiologist, contact the:

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